How can we address inequalities in access to genetic services?

CLINICAL GENETICS SOCIETY MEDICAL STUDENT PRIZE 2021-2022

Introduction

Genetic services involve discovering individuals whose genotype are associated with disease, and thus are at risk of developing the illness or passing on to their children (Law and Martin, 2020). This can have several benefits towards an individual's health through personalised medicine. For instance, Lynch Syndrome Screening can reduce morbidity and mortality from colorectal cancer from pharmacogenomic-based dosing of Warfarin to determine therapeutic dose, an anticoagulant used for the prevention of thromboembolic events (Institute of Medicine *et al.*, 2010). Alternatively, risk reduction interventions, such as lifestyle changes, early detection, medication, or surgery, may be available for those with genetic mutations associated with disease (Cancer Research UK, 2021). This holds promise for a future with public health genomics, where increasingly precise interventions target individual risk levels to provide cost-effective disease prevention, screening and surveillance programmes to ultimately benefit the population (Molster *et al.*, 2018).

However, there exist challenges in its utility. Genetic services are highly technical, involving multiple office visits, extensive medical assessment, and primary care referrals, thus are particularly vulnerable to accessibility barriers (Andrulis, 1998). Therefore, we shall be discussing the various barriers preventing equitable access to genetic testing, their causes, and possible solutions.

What are the Genetic Services available?

Genetic services offered in the UK entails genetic screening for various types of potentially hereditary diseases and cancers, personalised medicine, reproductive genomic sequencing, as well as genetic counselling. These are summarised in table 1:

	Adult Screening/Personalised Medicine	**Early/Reproductive Genomic Screening**	**Genetic Counselling**

Summary of service	• Genomics-led diagnosis of cancer to achieve earlier diagnosis and treatment
	  ○ E.g., Chronic Lymphocytic Leukaemia (CLL) has recognised subtypes defined by different prognosis and responses.
	  ○ E.g., Presence of DPYD gene mutation reduces metabolism of fluoropyrimidines used for cancers, resulting in severe adverse reaction.
	• Allowing for genomics-enabled clinical trials for cancer patients
	• Risk prediction tools used e.g., Polygenic Risk Scores (PRS) to allow for preventive strategies.
		• The new-born blood spot test tests for 9 rare but serious conditions where interventions are available before symptoms manifest.
		• Non-invasive Prenatal Testing (NIPT) detects aneuploidies such as Down Syndrome.
		• Pre-implantation Genetic Diagnosis (PGD) is an embryo test used to detect and avoid passing on serious genetic conditions.
			• A genetic counsellor can help patients to understand:
			  ○ Risk and benefit of test
			  ○ Meaning of potential results
			  ○ How family is affected
			  ○ Risk of passing to children
			  ○ Options for child and to avoid passing on hereditary diseases

When it is given	• Health condition may be genetic • Family member has health condition that is genetic • Close relatives have cancer that may be inherited	• Suspect a health condition may be passed on to children	• Patients may be referred to a genetic counsellor if offered a genetic test
How the screening results are used	• Understand whether an inherited health condition affects the patient, child or family member • Show risk of getting certain health conditions e.g., types of cancers • Guide doctors in providing medicine/treatment • Guide doctors on whether to patient can join clinical trials	• Diagnose rare health condition in child • Understand whether an inherited health condition affects the patient, child or family member • Show risk of getting certain health conditions e.g., types of cancers • Guide doctors in providing medicine/treatment • Guide doctors on whether to patient can join clinical trials	*NA*

Table 1 Summary Table of the different types of genetic services offered in the UK. Adapted from (NHS, 2019a; Department of Health & Social Care, 2020)

Why do inequalities in access exist?

Inequalities stem from vulnerable groups which are unaccounted for in the conduct of genetic services, such as the disabled community. Many who are deaf may miss relevant health information due to difficulty in translating concepts into sign language, thus they are less likely knowledgeable about genetic services and must work through an interpreter to communicate with healthcare professionals (Cooke-Hubley and Maddalena, 2011). This therefore presents major barriers in their access to genetic services.

Similarly, there exist social vulnerabilities that restrict one's access, such as socioeconomic status and ethnicity. Poor socioeconomic status can prevent the effective use of genetic services, despite the equalising role that NHS supposedly plays. While genetic testing by NHS is free when referred, inequalities can still exist given the "inverse equity law" where universal health services increases the health divide between the rich and poor (Sayani, 2019). These occur in populations of socioeconomic deprivation where they are unable to access as high a quality of healthcare service, as supported by various national surveys and data on screening and immunisation (Hart, 1971; Appleby and Deeming, 2001). This is compounded by the limited resources and lower levels of education (Yazbeck, 2009). Thus, the resulting difficulties in physical access from the former, such as transportation, and access to information in the latter, reduces the poor's ability or willingness to utilise health services (Yazbeck, 2009), including genomic testing. Akin to the deaf community, the disparity in access between the rich and poor are exacerbated by the knowledge-based disparity as accessibility is strongly associated to patients who are their own advocate (Hall and Olopade, 2005; Cooke-Hubley and Maddalena, 2011) and where patients who inquire about genetic testing are more likely to receive referrals (Wideroff *et al.*, 2003). On the other hand, those from a higher socioeconomic background are more likely informed and demanding of the health care system (Maddison, Asada and Urquhart, 2011) and thus better equipped to access genomic services. This therefore demonstrates how knowledge-based divide between the socioeconomic classes can result in unequal access to genetic testing.

Barriers within the wider healthcare system may be present as well. Yazbeck's adapted a framework (Figure 1) presenting further systemic barriers (summarised in Table 2) where multiple

steps are missing or inadequate for the deprived to effectively utilise genomic services (Yazbeck, 2009). Together, both the disparity in information and systemic barriers in health services lacking considerations for the deprived inexplicitly contribute to the growing differences in accessibility to genetic services.

Figure 1: Adopted from (Yazbeck, 2009) Eight Steps to Effective Use of Health Services by the Poor

Relevant Steps	***Issues***
2/3) Availability of human/material resources	• Shortages in supply of tests and genetic counsellors are more likely in deprived areas. A survey conducted in rural Illinois found that while majority of participants were aware of genetic testing, clinics nearby may not offer these services (46.9%) or that there are no accessible genetic counsellors with whom to discuss results (45.6%) (Fogleman *et al.*, 2019).
4) Organisational Quality	• Hours of operation are incompatible with working poor to leave work and seek services (Yazbeck, 2009) • Long waits at facilities increases loss incurred on the poor (Yazbeck, 2009). This is worsened by COVID-19 where hospitals were experiencing delays with genomic testing, as in Sheffield Diagnostic Genetic Service (Kirk, 2021) and poor reporting times of only 43% on target in Leeds Genetics Laboratory (The Leeds Teaching Hospital, 2019).
6) Timing and Continuity	• Delays faced in obtaining referrals and receiving genomic testing results may be detrimental in the patients seeking early treatment and prevention for severe genetic diseases.

Table 2: Summary of barriers to access to genetic health service for the Poor

Another major inequality is in ethnicity. There exists a racial disparity in use of genomic testing: in the US, Non-Hispanic Whites are more likely than Hispanic and Non-Hispanic Blacks in both receiving genetic tests and in discussing results with genetic counsellors (Carroll *et al.*, 2019) in the UK, ethnic minority are significantly under-represented among cancer genetic referrals (Mehta and Saggar, 2005), despite age-standardised incidence of cancer (excluding non-melanoma skin cancer) to be similar between White and Black males, at 0.41% to 0.42% for the former and 0.32% to 0.49% for the latter (National Cancer Intelligence Network and Cancer Research UK, 2009). Thus, there is a need to explore the relationship of genetics, culture and kinship patterns, as well as health beliefs towards both genetic screening and prenatal diagnosis (Mehta and Saggar, 2005). This difference can again be attributed to knowledge-based disparity (Hall and Olopade, 2006) where there exists a divide in education levels with regards to hereditary disease. A study in southeastern Georgia where groups least informed about breast cancer susceptibility genes are composed of African Americans (MacNew *et al.*, 2009). Moreover, sociocultural barriers to genetic information may be present. In Canada, 87% of genetic counsellors are white Caucasians who only speak English (Sayani, 2019). This poses a hurdle for non-English speaking patients with different religious and cultural beliefs (Sayani, 2019), resulting in difficulties communicating genetic concepts and understanding, which is further widening a

knowledge-based disparity. Accessibility is also hampered due to poor communication of family history or inaccurate risk perception, resulting in a significant underutilisation of genetic services, despite existing family history (Armstrong *et al.*, 2005).

This is worsened by ineffective risk-reducing measures post-genetic test and a deficit in ethnic diversity within genetic databanks (Hall and Olopade, 2006). Those from ethnic minorities may find it more difficult to enter clinical trials, where it was found that breast cancer prevention trials were more likely eligible for white women than Hispanic women, at a factor of 10, and black women, at a factor of 45 (Grann *et al.*, 2005). Moreover, majority of genetic data used to develop risk models are from high-risk white families, thus may not be generalisable to a non-white population (Hall and Olopade, 2006). As recently as 2017, genetic datasets are still composed of 88% with European descent (Mills and Rahal, 2019). This lack of accurate genetic predisposition data will hamper the ability for racial minorities to seek preventive measures post-screening. In extreme cases, a misdiagnosis may occur, flagging harmless genetic variants (Amsen, 2019). Thus, there is a degradation of trust in genetic services for those of different ethnicity (Amsen, 2019), inevitably exacerbating inaccessibility to genomic medicine.

However, it is also worth noting that not every racial difference is inequality. Differences in preference, where African American women were likely to make a decision to attend genetic counselling based on "personal and family values" (Halbert *et al.*, 2012), may suggest cultural differences instead.

How can we address these inequalities?

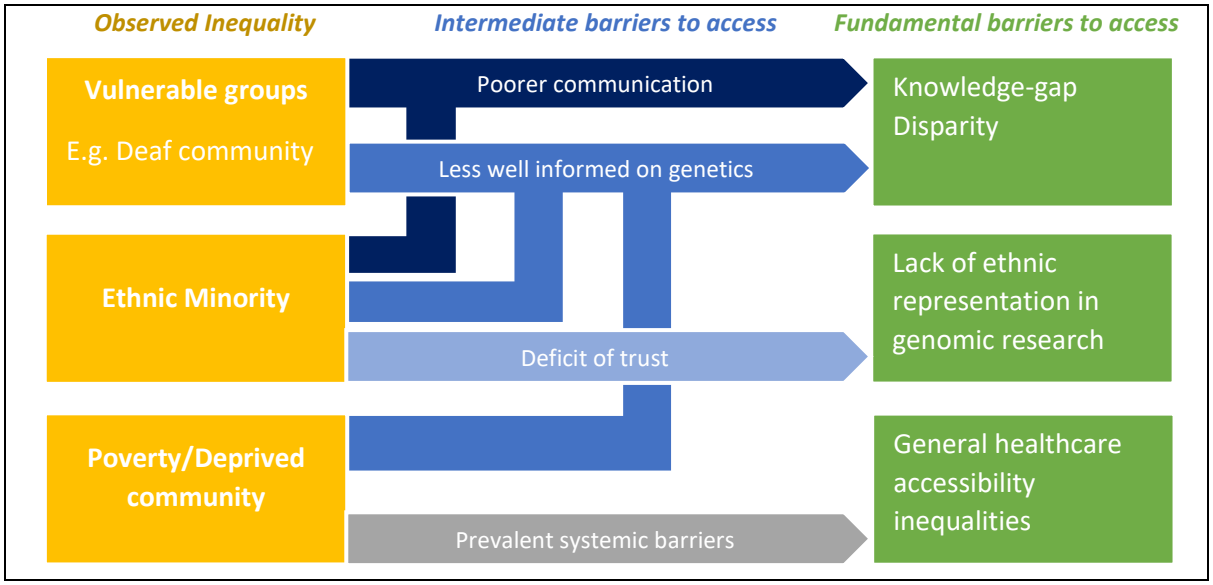

Figure 2: Summary of inequities in access and their associated causes

A cause common across access inequities is in the knowledge regarding the use of genetic services, whereby only the well informed would fully realise the benefits of genomic testing (Hall and Olopade, 2006). Public education on the mechanisms of genetics and hereditary disease risks should be communicated widely across disability, socioeconomic and racial barriers. This can be implemented in a similar fashion to other health promotion messages within specific contexts such as schools, hospitals, workplaces or residences (Kumar and Preetha, 2012). Only when patients are equally empowered to inquire about their genetic health will their access to genetic testing not be restricted (Wideroff *et al.*, 2003). However, several pitfalls in genomic education must be avoided. Information should avoid complex terminology or creating negative narratives which causes a fear of disease,

instead focusing on nuanced but simplified approaches (Skyers, 2018). Beyond general education, specific access barriers contributing to the knowledge-gap must be addressed as well. For instance, the communication of genetic information is hampered in both deaf communities (Cooke-Hubley and Maddalena, 2011) and in those who do not speak English (Sayani, 2019). Specific sign languages may be created to cater to communication with the deaf community. Additionally, provision of professional translators, preferably trained in genetics, should be available to communicate genetic information between these vulnerable groups and healthcare professionals, instead of a reliance on ad hoc translation (Mehta and Saggar, 2005). Importantly, improved accuracy in communication will reduce incorrect judgements on rationing of genetic services by the doctor due to misunderstanding family history or risk perception (Armstrong *et al.*, 2005). Therefore, steps taken to reduce the knowledge-inequality are critical in minimising subsequent barriers to access to genetic services.

Secondly, the quality of genetic services and research must be improved. This includes increasing diversity in ethnic participation within genomic research. Genomics England researchers identified organisational and attitudinal barriers with a lack of diversity in teams recruiting for 100,000 Genome project and within genetic counsellors (Skyers, 2018). This could easily result in unintentional bias, where judgments are made about community disinterest and lack of proactive engagement becomes pervasive (Skyers, 2018). Therefore, there is a need to set out regulatory NICE and commissioning pathways in line with Equality Act 2010, for both recruitment and clinical practice, and actively engage underserved communities on information and participation in genomic (Mehta and Saggar, 2005; Skyers, 2018). In summary, it is important to recognise racial disparity within genomics, and to address the gaps through proactive engagement of these communities. This can greatly improve the quality and trust in genetic services, thereby reducing the barriers in its accessibility.

Lastly, there is also a pressing need to address prevalent inequalities in access across the entire healthcare system as well. As emphasised in the NHS Long Term Plan, there is a need to improve universal access to healthcare to reduce health inequalities (NHS, 2019b). By acknowledging and creating specific measures towards vulnerable groups as identified in table 3, relevant barriers for each section of society can be broken down to equalise healthcare access. I believe this plan, if adhered to and appropriately funded, would be able to address many factors compounding health inequalities in the UK.

Chapter 2: Stronger NHS action on health inequalities
2.25: Targeted funding for locations with high health inequalities 2.26-2.39: Supporting local planning in health inequality reduction: • Targeted continuity of carer model in maternity services for vulnerable mothers and babies • Increase number of people receiving physical health checks to target higher risk of physical health on those suffering from mental illnesses • Investing in services and providing support for families where there is a child with learning disability to live happier, healthier, longer lives. • Investment to meet the healthcare needs of the homeless • Identify and support carers, such as out-of-hour options • Partner with local charities, social enterprise, and community interest companies in providing service and support to vulnerable and at-risk groups

Table 3: Summary of NHS Long term Plan (NHS, 2019b)

Conclusion

While there is visible progress in improving accessibility to genetic services, as seen through reports such as the Genetics White Paper (Department of Health, 2003) and genetic pilot programmes

(Bennett, Burton and Farndon, 2007), more can and must be done. Inequalities existing in all aspects of health should be addressed in addition to the wide knowledge-gap disparity, which presents a huge obstacle in accessing genetic services. Beyond public education, communication and translation specialists must be trained and recruited to improve the flow of information between patients and healthcare professionals. Additionally, it is important to proactively engage underserved communities, especially the ethnic minorities, to ensure results from genomic research are yielded inclusively. There is an urgent need to address these inequities, not only for population and individual health outcomes, but in frontier bioethics as well. It is beyond doubt that gene therapy can be beneficial, but it entails a myriad of ethical issues to disentangle (Roberts, 2010). However, as inequalities are intricately linked with ethical considerations (Lane, 2004), it is important to address them before we may adequately move forward ethically in cutting-edge genomic technology.

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